RSPO4 gene

R-spondin 4

Normal Function

The *RSPO4* gene provides instructions for making a protein called R-spondin-4. R-spondin-4 plays a role in the Wnt signaling pathway, a series of steps that affect the way cells and tissues develop. Wnt signaling is important for cell division, attachment of cells to one another (adhesion), cell movement (migration), and many other cellular activities. The role of R-spondin-4 is to increase Wnt signaling.

During early development, Wnt signaling plays a critical role in the growth and development of nails. R-spondin-4 is active in the skeleton and contributes to limb formation, particularly at the ends of the fingers and toes, where nail development occurs.

Different regions (domains) of R-spondin-4 have different functions; two regions known as furin-like domains are required for turning on (activating) and stabilizing proteins that play integral roles in the Wnt pathway.

Health Conditions Related to Genetic Changes

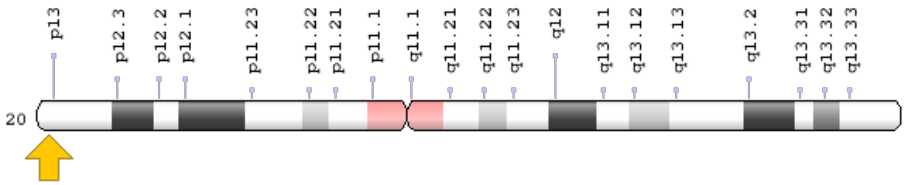
anonychia congenita

At least 17 mutations in the *RSPO4* gene have been found to cause anonychia congenita. This condition is characterized by the absence of fingernails and toenails (anonychia) from birth. Nearly all of the *RSPO4* gene mutations that cause this condition affect the furin-like domains, impairing protein function. Some mutations disrupt the structure of the furin-like domains, and others lead to the production of an abnormally short protein that does not contain these domains. As a result, R-spondin-4 cannot participate in the Wnt signaling pathway and nails develop improperly or not at all.

Chromosomal Location

Cytogenetic Location: 20p13, which is the short (p) arm of chromosome 20 at position 13

Molecular Location: base pairs 958,452 to 1,002,264 on chromosome 20 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- C20orf182
- CRISTIN4
- dJ824F16.3
- hRspo4
- R-spondin family, member 4
- roof plate-specific spondin-4

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): The Wnt Pathway https://www.ncbi.nlm.nih.gov/books/NBK10043/#A1061
- Madame Curie Bioscience: Secreted Antagonists/Modulators of Wnt Signaling https://www.ncbi.nlm.nih.gov/books/NBK6536/

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RSPO4%5BTIAB%5D%29+OR+%28R-spondin+4%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- R-SPONDIN FAMILY, MEMBER 4
  http://omim.org/entry/610573

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RSPO4%5Bgene%5D
- HGNC Gene Family: Endogenous ligands
  http://www.genenames.org/cgi-bin/genefamilies/set/542
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=16175
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/343637
- UniProt
  http://www.uniprot.org/uniprot/Q2I0M5

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